HEALTH AND SENIOR SERVICES

DIVISION OF FAMILY HEALTH SERVICES

Newborn Screening Program

Newborn Biochemical Screening

Proposed Amendments: N.J.A.C. 8:19-2.3 through 2.7 and 2.9

Authorized By: Clifton R. Lacy, M.D., Commissioner,

Department of Health and Senior Services

Authority: N.J.S.A. 26:2-110 and 26:2-111

Calendar Reference: See Summary below for explanation of exception to the Calendar Requirement.

Proposal Number: PRN 2003-362

Submit comments by June 3, 2004 to:

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Summary

Newborn biochemical screening is a public health activity aimed at the early identification of infants who are affected by metabolic disorders. Early identification of these conditions is particularly crucial, as timely intervention can lead to a significant reduction of morbidity, mortality and associated disabilities in affected infants. N.J.S.A. 2:26-110 and 111 specify that newborns will be tested for hypothyroidism, galactosemia, phenylketonuria, and other preventable biochemical disorders. The Commissioner of the New Jersey Department of Health and Senior Services (hereafter referred to as "the Commissioner") has the authority to require testing of newborn infants for other preventable biochemical disorders if reliable and efficient testing techniques are available. It is under this authority that the Commissioner is expanding newborn biochemical screening to include additional disorders.

Advances in screening technologies, coupled with public advocacy, led the New Jersey Department of Health and Senior Services (hereafter referred to as "the Department") to more closely examine the issue of newborn biochemical screening. In April 2000, then Commissioner Grant convened the Newborn Screening Advisory Panel. This panel was charged with reviewing newborn screening practice in New Jersey, reviewing additional disorders, and making recommendations concerning the appropriateness of mandated screening for additional biochemical disorders. The panel also heard open testimony from interested parties, including the public, during two public hearings. The panel presented its findings to the Commissioner in December 2000, recommending that newborn screening be expanded through the addition of 10 disorders to the current mandated four disorders.

New Jersey expanded biochemical screening of newborns to include 10 additional disorders: congenital adrenal hyperplasia, biotinidase deficiency, maple syrup urine disease, cystic fibrosis, medium chain acyl-CoA dehydrogenase (MCAD) deficiency, short chain acyl-CoA dehydrogenase (SCAD) deficiency, long chain acyl-CoA dehydrogenase (LCAD) deficiency, very long chain acyl-CoA dehydrogenase (VLCAD) deficiency, citrullinemia, and argininosuccinic acidemia. The first four of these disorders were added to the newborn screening panel in July 2001, and the remaining six disorders were added as of July 2002. Newborns continue to be tested for the four disorders included on the previous panel: phenylketonuria, congenital hypothyroidism, galactosemia, and hemoglobinopathies, including sickle cell anemia. N.J.A.C. 8:19-2, sets forth the requirements for newborn biochemical screening.

Subchapter 2 specifies the rules for the implementation of N.J.S.A. 26:2-110 and 111 (P.L. 1988, c.24), the statutory basis for New Jersey's Newborn Biochemical Screening Program. The Newborn Biochemical Screening Program is comprised of two State agencies, the Inborn Errors of Metabolism (IEM) Laboratory, and the Newborn Biochemical Screening (NBS) Follow-up Program. Newborns in New Jersey are presently tested for disorders which, if not treated very early in life, can cause mental retardation or death. Irreversible damage may occur even before there is any clinical evidence of a problem. The purpose of the Newborn Biochemical Screening Program is to identify affected infants through special tests, and assure that they receive appropriate treatment. The Newborn Biochemical Screening Program continues to be successful in identifying affected newborns early. Each year approximately 160 New Jersey newborns are saved from mental retardation, death, or serious illness because of early identification and timely referral for appropriate care. It is expected that through expanded screening, an additional 60 to

70 newborns will be identified with one of the disorders.

The rules describe the responsibilities of all the involved health care providers. Newborn screening for inborn errors of metabolism is a system that includes primary medical practitioners, hospital staff, laboratory personnel, administrative and follow-up personnel, tertiary care centers, specialists, and parents. This system must be designed to function smoothly and efficiently, insuring that all newborns are appropriately tested, retested (if needed), and followed if test results are found to be abnormal. For children with presumptive abnormal results, the NBS follow-up staff maintains communication with the appropriate physician to insure that established protocol is followed and appropriate treatment is received when indicated.

Since newborn screening is a system, staff from the IEM Laboratory and NBS Follow-up Program are in contact on a daily basis with hospitals, medical practitioners, and consultants. Additionally, usually semi-annually, formal meetings are held with medical consultant groups to discuss the current procedures and practices of the newborn screening system. During these meetings, the current law and rules are discussed. Additionally, staff from both programs conduct in-service training at hospitals to ensure all newborns are tested appropriately and that the hospital is in compliance with the law and rules.

Under the rules, hospitals have the primary responsibility for obtaining a satisfactory specimen. If the birth occurs outside of the hospital and the infant is not admitted to the hospital, the responsibility is that of the birth attendant. The proposed amendments: 1) specify, in N.J.A.C. 8:19-2.3 (c) and (d) the disorders to be added to mandated screening, as discussed above; 2) clarify, through specific reference, the paragraphs of N.J.A.C. 8:19-2.5, 2.6 and 2.7 that birth attendants, responsible physicians, and home health agencies must follow in submitting

specimens; 3) change the kind of report that the laboratory issues under N.J.A.C. 8:19-2.9 (a) 4 from "not clinically significant" to "within acceptable limits"; and 4) change delivery method hospitals use for specimens under N.J.A.C. 8:19-2.4 (a) 16 from first class mail to next day delivery.

As the Department has provided for a 60-day comment period for this notice of proposal, it is excepted from the rulemaking calendar requirement pursuant to N.J.A.C. 1:30-3.3 (b) 5.

Social Impact

The impact of a newborn screening program for inborn errors of metabolism is great, since it directly affects all newborns, parents of newborns, hospitals with maternity units, birthing centers, pediatric health care providers and the Department. Early identification and treatment of children with an inborn error of metabolism prevents the severe consequences of mental retardation or death. The positive social impact of the Newborn Biochemical Screening Program is clearly evident for the affected children, their families, and society as a whole.

To conduct biochemical screening, unless a parent objects for religious reasons, a small blood specimen (via a heel stick) is taken for analysis from every newborn in New Jersey and sent to the IEM Laboratory, Public Health and Environmental Laboratories, Department of Health and Senior Services. Nearly 125,000 specimens were received and assayed in 2001, of which nearly 11,000 were repeats because of some problem with the specimen or an abnormal test result. Infants with abnormal results reported by IEM are followed until the NBS Follow-up Program receives final case disposition or end-points are met. Medical specialists are available for

assistance with diagnosis and management.

The collection, handling, analysis and recordkeeping for over 114,000 specimens, the collection of requested repeats, the follow-up of all abnormal reports, the ongoing care of a cohort of identified children now numbering over 1,500, plus a supporting education program, presents a significant effort for the health care community, the Department and parents. The result of this effort has a demonstrable positive effect; early diagnosis and proper treatment for these affected children can make the difference between lifelong impairment and healthy development.

Changes in technology and improved testing procedures have had an affect on how newborns should be screened and followed. The social impact of the proposed amendments which add 10 disorders to the mandated screening panel as well as updating procedures and processes, reflecting current and best practices related to screening, testing, and follow-up procedures, will be positive.

Economic Impact

Newborn screening programs are effective in saving children from death or lifelong severe disability. The cost in dollars of early identification and treatment of affected infants is less than the cost of maintaining untreated children in institutions, special education programs and sheltered care settings. Before 1988, the newborn biochemical screening statute spoke only to early detection. The law was amended in 1988 to include both screening and the treatment of affected individuals. It directed the Commissioner of Health and Senior Services to ensure that treatment services are available to all identified, affected individuals. The amended law further requires that all revenues collected from the IEM fees be applied to the testing and treatment

procedures performed pursuant to the law. Hospitals and birthing centers collect and deliver the specimens to the IEM Laboratory. Costs for the testing, follow-up and treatment are covered by the fee charged for the IEM test. The IEM fee is specified in N.J.A.C. 8:45-2.1.

The proposed amendments to N.J.A.C. 8:19-2, Newborn Biochemical Screening, expand the number of disorders included in the screening, as well as clarifying collection procedures. Since there will be additional abnormal specimens, the entities responsible for the collection of initial and repeat specimens will have an increased work load. At this time, an increase in the IEM fee is expected due to the State's fiscal constraints, the exact increase in the cost of the laboratory, follow-up and treatment for expanded newborn screening has not yet been determined.

Federal Standards Statement

Currently, there are no Federal standards or requirements which mandate the testing of newborns for inborn errors of metabolism. Therefore, a Federal standards analysis is not required.

Jobs Impact

To conduct the biochemical blood test, a health care practitioner sticks the heel of the newborn and applies the blood to a special filter paper, which is then mailed to the Department. The test is performed as part of the routine care of newborns; as such, in hospitals, jobs are not generated or lost as a result of this rule. Expanded newborn screening will not affect the collection of specimens. However, to test the sample and follow-up newborns with abnormal results, the State employs staff in both the Inborn Errors of Metabolism Laboratory and the Newborn Biochemical

Screening Follow-up Program. The Division does not anticipate that there will be any impact on the generation or loss of jobs within the State resulting from the proposed amendments.

Agriculture Industry Statement

The proposed amendments will not have any impact on the agriculture industry.

Regulatory Flexibility Analysis

While these rules impose considerable reporting, recordkeeping and other compliance requirements, over 95 percent of the in-State births take place in hospitals, where the regulatory flexibility requirements are not applicable, since such hospitals employ more than 100 employees, and are not, therefore, small businesses as defined under the Regulatory Flexibility Act, N.J.S.A 52:14B-16 et seq. Less than one percent of births occur at home, in doctors' offices, or in the State's only birthing center, while the place of birth is not known (not stated on birth certificates) for the remaining births. The requirements of, and costs of compliance with, these amendments are discussed in the Summary and Economic Impact above. The Department believes that the requirements of this chapter must be uniformly applied in order to provide all newborns in the State, and their families, with the benefits of the programs. In the interest of public health, therefore, the Department does not believe that any exemption or differentiation should be made for non-hospital deliveries.

Smart Growth Impact

The proposed amendments will have no impact on the achievement of smart growth and the implementation of the State Development and Redevelopment Plan.

Full text of the proposal follows (additions indicated in boldface **thus;** deletions indicated in brackets **[thus]**):

SUBCHAPTER 2. NEWBORN BIOCHEMICAL SCREENING

8:19-2.3 Diseases and conditions tested

(a)-(b) (No change)

(c) Beginning July 2001, in addition to the disorders under (b) above, the following conditions were added to newborn screening:

1. **Maple syrup urine disease;**
2. **Congenital adrenal hyperplasia;**
3. **Cystic fibrosis; and**
4. **Biotinidase deficiency.**

(d) Beginning July 2002, in addition to the disorders under (b) and (c) above, the following conditions were added to newborn screening:

1. **Medium chain acyl-CoA dehydrogenase (MCAD) deficiency;**
2. **Short chain acyl-CoA dehydrogenase (SCAD) deficiency);**
3. **Long chain acyl-CoA dehydrogenase (LCAD) deficiency;**
4. **Very long chain acyl-CoA dehydrogenase (VLCAD) deficiency;**
5. **Citrullinemia; and**
6. **Argininosuccinic acidemia.**

8:19-2.4 Responsibilities of the chief executive officer

(a) The chief executive officer shall:

1.-15. (No change)

16. Assure that all specimens are forwarded to the testing laboratory within 24 hours of collection **[by first class mail or its equivalent]** **by next day delivery;**

17.-21. (No change)

8:19-2.5 Responsibilities of the birth attendant

(a) The birth attendant shall:

1. (No change)

2. Follow the specimen collection and sub-mission procedures specified in N.J.A.C. 8:19-2.4 **(a) 5 through 8 and 15 through 16;**

3.-4. (No change)

8:19-2.6 Responsibilities of the responsible physician

(a) The responsible physician shall:

1. (No change)

2. Comply with the specimen collection and submission procedures specified in N.J.A.C. 8:19-2.4 **(a) 5 through 8 and 15 through 16;**

3.-9. (No change)

8:19-2.7 Responsibilities of the home health agency

(a) The home health agency shall:

1. Follow the specimen collection procedures specified in N.J.A.C. 8:19- 2.4 **(a) 5 through 8 and 15 through 16;**

2.-3. (No change)

8:19-2.9 Responsibilities of the testing laboratory

(a) The testing laboratory shall:

1.-3. (No change)

4. Issue [reports of not clinically significant results] **result reports of within acceptable limits** to the chief executive officer or to the responsible physician, that is, the submitter of the specimen; and

5. (No change)